## APPENDIX "C"

- 1. Kosugi, S., et al (1993). Identification of thyroid-stimulating antibody-specific interaction sites in the N-terminal region of the thyrotropin receptor. Molecular Endocrinology, 7, 114-130.
- 5 2. Parma, J., et al (1993). Somatic mutations in the thyrotropin receptor gene cause hyperfunctioning thyroid adenomas. Nature, 365, 649-651.
  - 3. Duprez, L., et al (1994). Germline mutations in the thyrotropin receptor gene cause non-autoimmune autosomal dominant hyperthyroidism. Nature Genetics, 7, 396-401.
- 4. Matus-Leibovitch, N., et al (1995). Truncation of the thyrotropin-releasing hormone receptor carboxy tail causes constitutive activity and leads to impaired responsiveness in Xenopus oocytes and AtT20 cells. J. Biol. Chem., 270:3, 1041-1047.
  - 5. Kosugi, S., et al (1995). Characterization of heterogenous mutations causing constitutive activation of the luteinizing hormone receptor in familial male precocious puberty. Human Molecular Genetics, 4 (No.2), 183-188.
- 15 6. Magnusson, Y., et al (1994). Autoimmunity in idiopathic dilated cardiomyopathy. Circulation, 89, 2760-2767.
  - 7. Fu, M., et al (1994). Functional autoimmune epitope on alpha1-adrenergic receptors in patients with malignant hypertension. Lancet, 344, 1660-1663.
- 8. Samama, P., et al (1993a). A mutation-induced activation state of the B2-adrenergic receptor. J. Biol. Chem., 268:7, 4625-36.
  - 9. Kjelsberg, M.A., et al (1992). Constitutive activation of the alpha 1B-adrenergic receptor by all amino acid substitutions at a single site. J. Biol. Chem. 267, 1430-1433.
  - 10. Ren, Q., et al (1993). Constitutively active mutants of the alpha2-adrenergic receptor. J. Biol. Chem., 268, 16483-16487.
- 25 11. Burstein, E.S., et al (1996). Constitutive activation of chimeric m2/m5 muscarinic receptors and delineation of G-protein coupling selectivity domains. Biochem Pharmacol, 51:4, 539-44. Burstein, E.S., et al (1996). Amino acid side chains that define muscarinic receptor/G-protein coupling. Studies of the third intracellular loop. J. Biol. Chem., 271:6, 2882-5.
- Hasegawa, H., et al (1996). Two isoforms of the prostaglandin E receptor EP3 subtype different in agonist-independent constitutive activity. J. Biol. Chem., 271:4, 1857-1860.
  - 13. Nanevicz, T., et al (1996). Thrombin receptor activating mutations. J. Biol Chem., 271, 702-706.
  - 14. Boone, C., et al (1993). Mutations that alter the third cytoplasmic loop of the a-factor

- receptor lead to a constitutive and hypersensitive phenotype. Proc. Natl. Acad. Sci. (USA), 90:21, 9921-5.
- 15. Spiegel, A.M., et al (1995). Defects in G protein-coupled signal transduction in human disease. Ann. Rev. Physiol. 58, 143-170.
- 5 16. Seeman, P. (1993). Dopamine D4 receptors elevated in schizophrenia. Nature, 365, 441-445.
  - 17. Mann, J., et al (1986). Increased serotonin2 and beta-adrenergic receptor binding in the frontal cortices of suicide victims. Arch. Gen. Psychiat. 43, 954-959. Casey, C., et al (1996). Constitutively active mutant 5-HT2A serotonin receptors: inverse agonist activity of classical 5HT2A antagonists. Soc. Neurosci. Abstracts # 699.10
  - 18. Barker, E.L., et al (1994). Constitutively active 5-hydroxytryptamine2C receptors reveal novel inverse agonist activity of receptor ligands. J. Biol. Chem., 269:16, 11687-11690.
- 19. Maenhault, C., et al (1990). RCD8 codes for an adenosine A2 receptor with physiological constitutive activity. Biochem. Biophys. Res. Com., 173:3, 1169-1178.
  - 20. Parfitt, A.M., et al (1996). Hypercalcemia due to constitutive activity of the parathyroid hormone (PTH)/PTH-related peptide receptor: comparison with primary hyperparathyroidism. J. Clin. Endocr. Metab., 81, 3584-3588.
  - 21. Lavlie, R., et al (1996). The Ca(2+)-sensing receptor gene (PCAR1) mutation T151M in isolated autosomal dominant hypoparathyroidism. Hum Genet, 98:2, 129-33.
    - 22. Arvanitikis, L., et al (1997). Human herpesvirus KSHV encodes a constitutively active G-protein-coupled receptor linked to cell proliferation.
  - 23. Liu, J., et al (1996). Molecular mechanisms involved in muscarinic acetylcholine receptor-mediated G protein activation studied by insertion mutagenesis. J. Biol. Chem., 271:11, 6172-6178.
    - 24. Prezeau, L., et al (1996). Changes in the carboxy-terminal domain of metabotropic glutamate receptor 1 by alternate splicing generate receptors with differing agonist-independent activity. Mol. Pharmacol., 49, 422-429.
  - Alla, S.A., et al (1996). Extracellular domains of the bradykinin B2 receptor involved in ligand binding and agonist sensing defined by anti-peptide antibodies. 271, 1748-1755.
    - Wang, Z., et al (1994). Constitutive  $\mu$  opioid receptor activation as a regulatory mechanism underlying narcotic tolerance and dependance. Life Sciences, 54:22, 339-350.
  - Tiberi, M. & Caron, M.G. (1994). High agonist-independent activity is a distinguishing feature of the dopamine D1B receptor subtype. The J. Biol. Chem. 269:45. 27925-

10

27931.

- 28. Robbins, L.S., et al (1993). Pigmentation phenotypes of variant extension locus alleles result from point mutations that alter MSH receptor function. Cell, 72, 827-834.
- Eggerick, D., et al (1995). Molecular cloning of an orphan G-protein-coupled receptor that constitutively activates adenylate cyclase. Biochem. J. 389, 837-843.
  - 30. Jakub/Eik, J., et al (1995). Constitutive activity of the M1-M4 subtypes of muscarinic receptors in transfeceted CHO cells and of muscarinic receptors in the heart cells revealed by negative antagonists. FEBS Lett, 377:2, 275-9.
- 31. Burstein, E.S., et al (1995). Constitutive activation of muscarinic receptors by the G-protein Gq. FEBS Lett, 363:3, 261-3.
  - 32. Ter Lack, A., et al (1995). Modelling and mutation studies on the histamine H1-receptor agonist binding site reveal different binding modes for H1-agonisis: Asp116 (TM3) has a constitutive role in receptor stimulation. J. Computer-aided molecular design, 9, 319-330.